Stanford

Hetanshi Naik

Associate Professor (Teaching) of Genetics

Bio

BIO

Hetanshi Naik is an Associate Professor in the Department of Genetics and the Research Director of the MS Program in Human Genetics and Genetic Counseling. She is a board certified genetic counselor and clinical researcher with clinical expertise in the inborn errors of heme biosynthesis, the Porphyrias, lysosomal storage disorders (LSDs), and pharmacogenomics, and research expertise in clinical trials, patient reported outcomes (PROs), qualitative methods, and study design.

Her research interests include developing and evaluating PROs for genetic disorders and genomics, in particular assessing PROs as outcomes for clinical trials, pharmacogenomics implementation, and genetic counseling education and processes, as well as utilizing digital health technologies to improve clinical care, genetic counseling, patient reporting, trial efficacy, and outcomes.

ACADEMIC APPOINTMENTS

- Associate Professor (Teaching), Genetics
- Member, Maternal & Child Health Research Institute (MCHRI)

ADMINISTRATIVE APPOINTMENTS

- Research Director, MS Program in Human Genetics and Genetic Counseling, Stanford University School of Medicine, (2022- present)

BOARDS, ADVISORY COMMITTEES, PROFESSIONAL ORGANIZATIONS

- Research Faculty Working Group, Association of Genetic Counseling Program Directors (2022 - present)
- Board of Directors, United Porphyrias Association (2021 - present)
- Diversity Committee, Rare Diseases Clinical Research Network (2021 - present)
- Member, Rare Disease Diversity Coalition (2021 - present)
- Data Standards Committee, Rare Diseases Clinical Research Network (2020 - present)
- Founding Member, American Porphyrias Expert Collaborative (APEX) (2020 - present)

PROFESSIONAL EDUCATION

- PhD, Icahn School of Medicine at Mount Sinai , Clinical Research (2018)
- MS, Mount Sinai School of Medicine , Genetic Counseling (2010)
- BSc, McMaster University , Biology major, Genetics specialization (2008)

Teaching

COURSES

2022-23

- Genetic Counseling Research Seminar: GENE 282A (Win)

Publications

PUBLICATIONS

- **A pilot study of oral iron therapy in erythropoietic protoporphyria and X-linked protoporphyria.** *Molecular genetics and metabolism reports*
  Balwani, M., Naik, H., Overbey, J. R., Bonkovsky, H. L., Bissell, D. M., Wang, B., Phillips, J. D., Desnick, R. J., Anderson, K. E.
  2022; 33: 100939

- **Prospective observational pilot study of quantitative light dosimetry in erythropoietic protoporphyria.** *Journal of the American Academy of Dermatology*
  Dickey, A., Rebeiz, L., Raef, H., Leaf, R. K., Elmariah, S., Naik, H., Anderson, K., Conley, J., Iyasere, C., Zhao, S., Birkenfeld, J. S., Arroyo-Gallego, T., Wheeden, et al
  2022

- **Evidence-based consensus guidelines for the diagnosis and management of erythropoietic protoporphyria and X-linked protoporphyria.** *Journal of the American Academy of Dermatology*
  Dickey, A. K., Naik, H., Keel, S. B., Levy, C., Beaven, S. W., Elmariah, S. B., Erwin, A. L., Goddu, R. J., Hedstrom, K., Leaf, R. K., Kazamel, M., Mazepa, M., Philpotts, et al
  2022

- **Attitudes on pharmacogenomic results as secondary findings among medical geneticists.** *Pharmacogenetics and genomics*
  Bartos, M. N., Scott, S. A., Jabs, E. W., Naik, H.
  2022

- **Hepatocellular Carcinoma in Acute Hepatic Porphyrias: Results from the Longitudinal Study of the U.S. Porphyrias Consortium.** *Hepatology (Baltimore, Md.)*
  Saberi, B., Naik, H., Overbey, J. R., Erwin, A. L., Anderson, K. E., Bissell, D. M., Bonkovsky, H. L., Phillips, J. D., Wang, B., K Singal, A., M McGuire, B., Desnick, R. J., Balwani, et al
  2021; 73 (5): 1736-1746

- **Pharmacogenomic education among genetic counseling training programs in North America.** *Journal of genetic counseling*
  Loudon, E., Scott, S. A., Rigobello, R., Scott, E. R., Zinberg, R., Naik, H.
  2021

- **Experiences from the epicenter: Professional impact of the COVID-19 pandemic on genetic counselors in New York.** *American journal of medical genetics. Part C, Seminars in medical genetics*
  Bergstrom, K. L., Brander, T. E., Breen, K. E., Naik, H.
  2021; 187 (1): 28-36

- **Evidence in the UK Biobank for the underdiagnosis of erythropoietic protoporphyria.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Dickey, A. K., Quick, C., Ducamp, S., Zhu, Z., Feng, Y. A., Naik, H., Balwani, M., Anderson, K. E., Lin, X., Phillips, J. E., Rebeiz, L., Bonkovsky, H. L., McGuire, et al
  2021; 23 (1): 140-148

- **Gaucher disease and SARS-CoV-2 infection: Experience from 181 patients in New York.** *Molecular genetics and metabolism*
  Fierro, L., Nesheiwat, N., Naik, H., Narayanan, P., Mistry, P. K., Balwani, M.
  2021; 132 (1): 44-48

- **Digital Health Applications for Pharmacogenetic Clinical Trials.** *Genes*
  Naik, H., Palaniappan, L., Ashley, E. A., Scott, S. A.
  2020; 11 (11)

- **EXPLORE: A Prospective, Multinational, Natural History Study of Patients with Acute Hepatic Porphyria with Recurrent Attacks.** *Hepatology (Baltimore, Md.)*
  Gouya, L., Ventura, P., Balwani, M., Bissell, D. M., Rees, D. C., Stölzel, U., Phillips, J. D., Kauppinen, R., Langendonk, J. G., Desnick, R. J., Deybach, J. C., Bonkovsky, H. L., Parker, et al
  2020; 71 (5): 1546-1558

- **Evaluating the Patient-Reported Outcomes Measurement Information System scales in acute intermittent porphyria.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Naik, H., Overbey, J. R., Montgomery, G. H., Winkel, G., Balwani, M., Anderson, K. E., Bissell, D. M., Bonkovsky, H. L., Phillips, J. D., Wang, B., McGuire, B., Keel, S., Levy, et al
  2020; 22 (3): 590-597

- **The N370S/R496H genotype in type 1 Gaucher disease - Natural history and implications for pre symptomatic diagnosis and counseling.** *Molecular genetics and metabolism reports*
  Zeid, N., Stauffer, C., Yang, A., Naik, H., Fierro, L., Ganesh, J., Balwani, M.
  2020; 22: 100567

- **Knowledge and attitudes of Parkinson's disease risk in the Gaucher population.** *Journal of genetic counseling*
  Zaretsky, L., Zeid, N., Naik, H., Alcalay, R. N., Balwani, M.
  2020; 29 (1): 105-111

- **Knowledge and attitudes on pharmacogenetics among pediatricians.** *Journal of human genetics*
  Rahawi, S. n., Naik, H. n., Blake, K. V., Owusu Obeng, A. n., Wasserman, R. M., Seki, Y. n., Funanage, V. L., Oishi, K. n., Scott, S. A.
  2020; 65 (5): 437–44

- **Implementing a pharmacogenetic-driven algorithm to guide dual antiplatelet therapy (DAPT) in Caribbean Hispanics: protocol for a non-randomised clinical trial.** *BMJ open*
  Hernandez-Suarez, D. F., Melin, K. n., Marin-Maldonado, F. n., Nunez, H. J., Gonzalez, A. F., Gonzalez-Sepulveda, L. n., Rivas-Tumanyan, S. n., Naik, H. n., Ruaño, G. n., Scott, S. A., Duconge, J. n.
  2020; 10 (8): e038936

- **Clinical Pharmacogenetic Testing and the Posttest Counseling Conundrum.** *Clinical pharmacology and therapeutics*
  Rigobello, R. n., Rahawi, S. n., Wallsten, R. n., Cody, N. n., Nicoletti, P. n., Owusu Obeng, A. n., Naik, H. n., Dillon, M. W., Scott, S. A.
  2020; 108 (5): 924–28

- **Evaluating quality of life tools in North American patients with erythropoietic protoporphyria and X-linked protoporphyria.** *JIMD reports*
  Naik, H., Overbey, J. R., Desnick, R. J., Anderson, K. E., Bissell, D. M., Bloomer, J., Bonkovsky, H. L., Phillips, J. D., Wang, B., Singal, A., Balwani, M.
  2019; 50 (1): 9-19

- **Psychosocial issues in erythropoietic protoporphyria - the perspective of parents, children, and young adults: A qualitative study.** *Molecular genetics and metabolism*
  Naik, H., Shenbagam, S., Go, A. M., Balwani, M.
  2019; 128 (3): 314-319

- **Diagnostic Delay in Erythropoietic Protoporphyria.** *The Journal of pediatrics*
  Lala, S. M., Naik, H., Balwani, M.
  2018; 202: 320-323.e2

- **Parkinson's disease prevalence in Fabry disease: A survey study.** *Molecular genetics and metabolism reports*
  Wise, A. H., Yang, A., Naik, H., Stauffer, C., Zeid, N., Liong, C., Balwani, M., Desnick, R. J., Alcalay, R. N.
  2018; 14: 27-30

- **Clinical, Biochemical, and Genetic Characterization of North American Patients With Erythropoietic Protoporphyria and X-linked Protoporphyria.** *JAMA dermatology*
  Balwani, M., Naik, H., Anderson, K. E., Bissell, D. M., Bloomer, J., Bonkovsky, H. L., Phillips, J. D., Overbey, J. R., Wang, B., Singal, A. K., Liu, L. U., Desnick, R. J.
  2017; 153 (8): 789-796

- **Acute Intermittent Porphyria in children: A case report and review of the literature.** *Molecular genetics and metabolism*
  Balwani, M., Singh, P., Seth, A., Debnath, E. M., Naik, H., Doheny, D., Chen, B., Yasuda, M., Desnick, R. J.
  2016; 119 (4): 295-299

- **Experiences and concerns of patients with recurrent attacks of acute hepatic porphyria: A qualitative study.** *Molecular genetics and metabolism*
  Naik, H., Stoecker, M., Sanderson, S. C., Balwani, M., Desnick, R. J.
  2016; 119 (3): 278-283

- **The D519G Polymorphism of Glyceronephosphate O-Acyltransferase Is a Risk Factor for Familial Porphyria Cutanea Tarda.** *PloS one*
  Farrell, C. P., Overbey, J. R., Naik, H., Nance, D., McLaren, G. D., McLaren, C. E., Zhou, L., Desnick, R. J., Parker, C. J., Phillips, J. D.
  2016; 11 (9): e0163322

- **Pitfalls in Erythrocyte Protoporphyrin Measurement for Diagnosis and Monitoring of Protoporphyrias.** *Clinical chemistry*
  Gou, E. W., Balwani, M., Bissell, D. M., Bloomer, J. R., Bonkovsky, H. L., Desnick, R. J., Naik, H., Phillips, J. D., Singal, A. K., Wang, B., Keel, S., Anderson, K. E.
  2015; 61 (12): 1453-6

- **Afamelanotide for Erythropoietic Protoporphyria.** *The New England journal of medicine*
  Langendonk, J. G., Balwani, M., Anderson, K. E., Bonkovsky, H. L., Anstey, A. V., Bissell, D. M., Bloomer, J., Edwards, C., Neumann, N. J., Parker, C., Phillips, J. D., Lim, H. W., Hamzavi, et al
  2015; 373 (1): 48-59

- **Liver Transplantation for Acute Intermittent Porphyria: Biochemical and Pathologic Studies of the Explanted Liver.** *Molecular medicine (Cambridge, Mass.)*
  Yasuda, M., Erwin, A. L., Liu, L. U., Balwani, M., Chen, B., Kadirvel, S., Gan, L., Fiel, M. I., Gordon, R. E., Yu, C., Clavero, S., Arvelakis, A., Naik, et al
  2015; 21: 487-95

- **Acute porphyrias in the USA: features of 108 subjects from porphyrias consortium.** *The American journal of medicine*
  Bonkovsky, H. L., Maddukuri, V. C., Yazici, C., Anderson, K. E., Bissell, D. M., Bloomer, J. R., Phillips, J. D., Naik, H., Peter, I., Baillargeon, G., Bossi, K., Gandolfo, L., Light, et al
  2014; 127 (12): 1233-41

- **An uncommon 3.4-Mb interstitial deletion at 3q29.** *Clinical dysmorphology*
  Wang, J. C., Naik, H., Khan, A., Nowaczyk, M. J.
  2010; 19 (3): 133-136